

Opis choroby *

Definicja

A rare form of mucopolysaccharidosis characterized by abnormal storage of hyaluronan in lysosomes due to deficiency of hyaluronidase 1. Clinical manifestations include knee and/or hip pain associated with swelling, diffuse joint involvement with proliferative synovitis and occurrence of multiple periarticular soft-tissue masses, short stature, and dysmorphic craniofacial features (such as flattened nasal bridge, bifid uvula, and cleft palate).

Dane

Klasyfikacja	Synonimy
Choroba	MPS9 MPS9 MPSIX Mukopolisacharydoza typu 9 Mukopolisacharydoza typu IX MPSIX Mucopolysaccharidosis type 9 Mucopolysaccharidosis type IX

Kod ORPHA	Kod OMIM	Kod ICD10
67041	601492	E76.2

Kod ICD11
5C56.3Y

[*Źródło](#)

orphanet